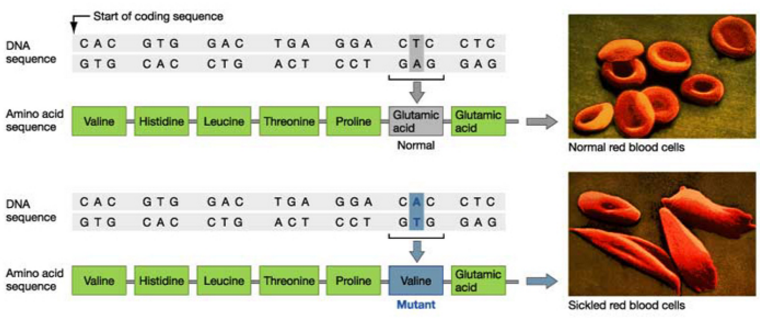
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**Mendelian Genetics Sickle Cell Anemia Analysis – Part 1**

Thank you to Ms. Glick

**Introduction:**

Hemoglobin is a protein found in red blood cells (RBCs) that transports oxygen throughout the body. The hemoglobin protein consists of four polypeptide chains: two alpha chains and two beta chains. Sickle cell disease (also called sickle cell anemia) is caused by a genetic mutation in the DNA sequence that codes for the beta chain of the hemoglobin protein. The mutation causes an amino acid substitution, replacing glutamic acid with valine. Due to this change in amino acid sequence, the hemoglobin tends to precipitate (or clump together) within the RBC after releasing its oxygen. This clumping causes the RBC to assume an abnormal “sickled” shape.



Individuals who are homozygous for the normal hemoglobin allele (HbA) receive a normal hemoglobin allele from each parent and are designated AA. People who are homozygous for normal hemoglobin do not have any sickled RBCs. Individuals who receive one normal hemoglobin allele from one parent and one mutant hemoglobin, or sickle cell allele (HbS), from the other parent are heterozygous and are said to have sickle cell trait. Their genotype is AS. Heterozygous individuals produce both normal and mutant hemoglobin proteins, thus it displays a codominance pattern of inheritance. These individuals do not have sickle cell disease, and most of their RBCs are normal. However, due to having one copy of the sickle cell allele, these individuals do show some sickling of their RBCs in low-oxygen environments. People with sickle cell disease are homozygous for the sickle cell allele (SS genotype); they have received one copy of the mutant hemoglobin allele from each parent. The resulting abnormal, sickle-shaped RBCs in these people block blood flow in blood vessels, causing pain, serious infections, and organ damage.

**Genetics Problems:**

1. If two people with sickle cell trait have children, what is the chance that a child will have normal RBCs in both high-and low-oxygen environments? What is the chance that a child will have sickle cell disease? Write the possible genotypes in the Punnett square.
   1. Normal RBCs in high- and low-oxygen environments (fraction of offspring) \_\_\_\_\_\_\_\_\_\_\_\_
   2. Sickle cell disease (fraction of offspring) \_\_\_\_\_\_\_\_\_\_\_\_



* 1. What is the chance that a child will carry the HbS gene but not have sickle cell disease? Express your answer as a fraction.
  2. What are the chances that these parents will have three children who are homozygous for normal RBCs? Express your answer as a fraction. (Show your work.)
  3. What are the chances that these parents will have two children with sickle cell trait and one with sickle cell disease? Express your answer as a fraction. (Show your work.)

1. A woman with sickle cell disease has children with a man who has sickle cell trait. Answer the following questions.
   1. What are the genotypes of the parents?
   2. What is the genetic makeup of the gametes the mother can produce? In other words, what alleles can she give to the gametes she creates?
   3. What is the genetic makeup of the gametes the father can produce? In other words, what alleles can he give to the gametes he creates?
   4. In the Punnett square, show all the possible genotypes of their children. Then summarize the genotype and phenotype frequencies of the possible offspring to the right as percentages.



* 1. What are the chances that any one of this couple’s children will have sickle cell disease? Express your answer as a percent.
  2. If this couple moves to the lowlands of East Africa and has children, which of their children would be more likely to survive? Explain your answer.